Statutory Approvals Committee – minutes

Centre 0101 (CARE Nottingham)

Pre-implantation Genetic Diagnosis (PGD) application for Netherton Syndrome, OMIM #256500

Thursday, 24 September 2020

HFEA, 10 Spring Gardens, London, SW1A 2BU via Teleconference

Committee members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr Jenny Carmichael	
Legal Adviser	Dawn Brathwaite	Mills & Reeve – LLP
Observers	Dee Knoyle Matthew Mudford (Induction) Cora Sweet (Induction)	Committee Officer Scientific Policy Officer Policy Officer

Declarations of interest:

- Members of the committee declared that they had no conflicts of interest in relation to this item.

The committee had before it:

- 9th edition of the HFEA Code of Practice.
- Standard licensing and approvals pack for committee members.

The following papers were considered by the committee:

- Executive Summary
- PGD Application Form
- Redacted Peer Review.

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Jenny Carmichael, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Netherton Syndrome, OMIM #256500, is consistent with the peer review.

1.3. The committee noted that the condition being applied for is not on the list of approved PGD conditions.

1.4. The committee noted that a Genetic Alliance UK statement had not been provided for this application.

1.5. The committee had regard to its decision tree. The committee noted that the centre is licensed to carry out PGD. The committee was also satisfied that the centre has experience of carrying out PGD and that generic patient information about its PGD programme and associated consent forms had previously been received by the HFEA.

1.6. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of Schedule 2 of the Act, i.e. 'where there is a particular risk that the embryo may have any gene, chromosome or mitochondrion abnormality, establishing whether it has that abnormality or any other gene, chromosome or mitochondrion abnormality'.

1.7. The committee noted that Netherton Syndrome, OMIM #256500, is inherited in an autosomal recessive manner, which means there is a 25% chance of an embryo being affected by the condition in each pregnancy if each parent has a relevant mutation.

1.8. The committee noted that the penetrance of the condition is 100%.

1.9. Netherton Syndrome, OMIM #256500 is characterised by red, scaly skin and abnormal hair shafts. Infants are at risk of becoming dehydrated and developing respiratory infections, emphysema, and sepsis, which can be life threatening. Death can occur within the first days of life. Throughout childhood and into adulthood, affected individuals continue to be prone to infections. Seizures and developmental delay, with intellectual disabilities and muscular abnormalities are common.

1.10. There is no cure for this condition and treatment focuses on managing the symptoms of patients.

1.11. The committee noted the executive's request to consider Netherton Syndrome, OMIM #256500, for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that, in the worst-case scenario, Netherton Syndrome, OMIM #256500, is a rare, and potentially life-limiting condition with symptoms present at birth. There is no cure for the condition which can cause a high degree of pain and suffering for those affected and can lead to death in the neonatal period or within the first few years of life. The committee considered the potentially devastating physical and psychological impact on the quality of life of those affected with the condition.

2.2. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk that a person with such an abnormality will, given the condition's worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

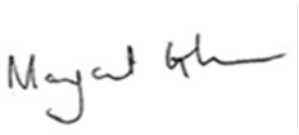
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Netherton Syndrome, OMIM #256500

3. Chairs signature

3.1. I confirm this is a true and accurate record of the meeting.

Signature



Name
Margaret Gilmore

Date
20 October 2020